Sickle cell anemia detected in fetuses

For the first time, two independent groups of researchers have successfuly managed to diagnose sickle cell anemia in living human fetuses. Although the technique is still highly experimental and risky, it will probably eventually become available to the general population, and then it could have a widespread and profound impact on the black community, which is largely, although not exclusively, afflicted with the disease. About 10 percent of American blacks carry a gene for sickled hemoglobin. About one out of every 500 American blacks actually has the disease (carries two genes for sickled hemoglobin). Those with the disease, not the carriers, would be the target of the fetal diagnostic procedure.

The researchers include Yuet Wai Kan, Mitchell S. Golbus and Richard Trecartin of the University of California at San Francisco and Blanche P. Alter, Shlomo Friedman, John C. Hobbins, Maurice J. Mahoney, Anita S. Sherman, John F. McSweeney, Elias Schwartz and David G. Nathan of Children's Hospital Medical Center and Harvard Medical School in Boston, Children's Hospital of Philadelphia and Yale-New Haven Hospital. Both groups report their results in the May 6 NEW ENGLAND JOURNAL OF MEDICINE.

Successfully diagnosing the fetus for sickle cell anemia first requires obtaining blood from the fetus without disturbing the pregnancy. The technique used by Kan's group consists of scanning the pregnant woman with ultrasound to locate the fetus, inserting a long needle into the amniotic cavity around the fetus, and positioning the needle so that one is able to withdraw a specimen of blood only. The technique used by Alter's group makes use of a fetuscope, a fiberoptic needle instrument through which the obstetrician can look at the fetus and visualize a fetal blood vessel. The instrument has a side arm hole through which the obstetrician brings down another needle. He can thus see himself putting the needle into the fetal blood vessel. "Hobbins is the only obstetrician who has used this technique successfully for getting fetal blood in ongoing pregnancies," Alter explains.

Then comes the challenge of taking the tiny drop of withdrawn fetal blood and incubating it with radioactive amino acids. The fetal blood uses radioactively labeled amino acids to make new hemoglobin chains. The hemoglobin chains are then submitted to chromatography to determine whether there are sickled beta chains present—that is, whether the fetus has the disease. The radioactive labeling is necessary because there is so little fetal blood present. Radioactivity also offers a selective advantage for revealing fetal chains if fetal blood is contaminated with maternal blood. This is because the mother's blood doesn't incorporate amino acids into new hemoglobin chains as rapidly as the fetal blood does.

These techniques were conducted on fetuses 20 weeks old, about the upper limit for therapeutic abortions.

Where do the two groups go from here? Alter says her group is going to work on "improvement of methods for obtaining fetal blood, detecting what the fetus is synthesizing and getting some of the bugs out of the system in both obstetrics and hematology." According to Golbus, the San Francisco group "will continue to offer it to couples who are at risk for having a sickle-cell baby, meanwhile trying to perfect techniques of safely obtaining blood and using less and less blood. But I don't think that's the final answer. I think we'll eventually find some way of getting the answer out of amniotic fluid. But that's 10 years off at least."

Fetal diagnosis of sickle cell anemia, like that of some other diseases, raises ethical considerations. There is as yet no way to treat diseased fetuses in the womb. If a fetus is found to have the disease, the only recourse is to let it be born diseased or to abort it. So should fetuses diagnosed for the disease always be aborted? "There is no such thing 'as in all cases,'" Golbus replies. "Different couples will handle the information differently, and they should."

Alter, however, takes a somewhat different stance. Since the test is risky, she doesn't believe it should be undertaken unless a couple has already decided on an abortion if their fetus has the disease. □

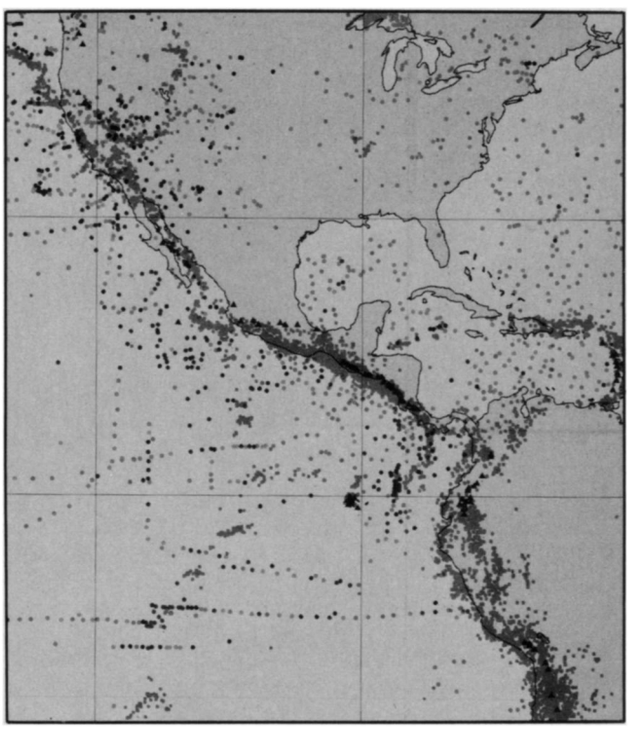
FermiLab achieves 500 GeV protons

Physicists at the Fermi National Accelerator Laboratory near Batavia, Ill., last week produced the first 500 billion-electron-volt (GeV) protons in a manmade machine—the half-way mark in their efforts to explore the nucleus with particles accelerated to one tera-electron-volts (1 TeV = 1000 GeV). The achievement came as the result of modifications made on the existing accelerator—originally designed to produce 200 GeV particles—and later addition of superconducting magnets will allow doubling to 1 TeV (SN: 1/4/75, p. 11).

Robert C. Seamans Jr., head of the Energy Research and Development Administration (ERDA) made the announcement, saying "This achievement maintains the position of the FermiLab accelerator as the highest energy machine in the world." The new record was set early on the morning of May 14 and surpasses an old record of 450 GeV, set at the lab last August.

The increased energy was made possible by several improvements in the power supply system, including installation of a new primary power transformer, a modified energy storage system, new high-voltage lines and improvement in cooling the magnets. FermiLab is operated for ERDA by a consortium of 52 universities in the United States and Canada and achieved its design energy of 200 GeV in March, 1972. □

Mapping the world's heat flow



NOAA

World heat flow map, showing the amount of heat flowing outward from the earth's interior (indicated by differently shaded dots) together with active volcano sites (triangles) and earthquake epicenters (squares), has been produced by the National Oceanic and Atmospheric Administration's World Data Center A for Solid Earth Geophysics in Boulder, Colo. North-South American segment (shown) indicates only a portion of the 5,500 global heat-flow measurements, representing several decades of study.